CURRICULUM VITAE of JOAN H.M. KNOLL

Education:

1979 B.Sc. (Biology, Genetics) 1979, University of Western Ontario
1982 M.Sc. (Genetics) 1982, University of Saskatchewan, Saskatoon
1987 Ph.D. (Human Genetics) 1987, University of Manitoba

Postdoctoral Training:
Research Fellowship:

1987-90 Research Fellow in Medicine (Genetics), Children's Hospital, Boston
1987-90 Fellow in Pediatrics, Harvard Medical School

Certification:

1990 Board Certified in Clinical Cytogenetics, American Board of Medical Genetics
1992 Board Certified in Clinical Cytogenetics, Canadian College of Medical Genetics
1992 Fellow, Canadian College of Medical Genetics
1993 Founding Fellow in American College of Medical Genetics
1996 Board Certified in Clinical Molecular Genetics, American College of Medical Genetics

Academic Appointments:

1990 Instructor in Pediatrics, Harvard Medical School
1991 Instructor in Pathology, Harvard Medical School
1992-99 Assistant Professor in Pathology, Harvard Medical School
1999- Associate Professor in Medicine/Pediatrics, UMKC School of Medicine

Hospital Appointments:

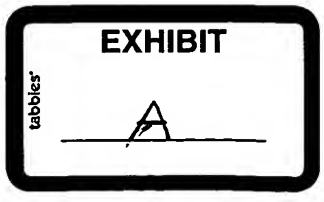
1990-1999 Research Associate in Medicine (Genetics), Children's Hospital, Boston
1991-1998 Co-director, Clinical Cytogenetics Service, Beth Israel Deaconess Medical Center, Boston
1998-1999 Director, Clinical Cytogenetics Service, Beth Israel Deaconess Medical Center, Boston
1999- Director, Clinical Cytogenetics Service, Children's Mercy Hospital, Kansas City
1999- Professorship in Pediatric Cytogenetics, Children's Mercy Hospital

Major Committee Assignments:
National and Regional:

1991-1995 Scientific Advisor, Angelman Syndrome Foundation
1992-present Scientific Advisor, Canadian Angelman Syndrome Society
1994-2000 Scientific Advisor, Scientific Advisor, Inverted Duplication 15 Education and Advocacy Group
1994 Member, Technology Transfer Subcommittee for diagnostic testing of Angelman and Prader-Willi syndromes, American College of Medical Genetics
1996 Co-organizer of Third International Chromosome 15 Workshop in October (Vancouver)

Memberships and Committee Assignments in Professional Societies:

1986- present American Society of Human Genetics
1989- present Reviewer for Genetics journals (American Journal of Human Genetics, American Journal of Medical Genetics, Cancer Genetics and Cytogenetics, Cytogenetics and Cell Genetics, Genomics, Human Molecular Genetics, New England Journal of Medicine)
1991-97 Member, Board of Directors, Angelman Syndrome Foundation
1992- Fellow in Canadian College of Medical Genetics
1992- Canadian Angelman Syndrome Society
1993- Founding Fellow in American College of Medical Genetics
1993-1999 Member, Prenatal Diagnosis Committee of New England Genetics Group
1993-99 Cytogenetics Faculty Organizer and Member, Harvard Training Fellowship Program in Genetics
1994- Member, Inverted Duplication 15 Education and Advocacy Group
2000- present American Cancer Research Association, Active Member



Major Research Interests:

1. Genome organization and Chromatin/chromosomal structure
2. Genetic imprinting
3. Phenotype/genotype relationships of loci within 15q11q13, including Angelman syndrome, Prader-Willi syndrome and duplication syndromes
4. Human disorders with chromosomal rearrangements

Principal Clinical and Hospital Service Responsibilities:

1991-1999 Scientific Director, Clinical Cytogenetics Service, Beth Israel Hospital, Boston
1999- Director, Clinical Cytogenetics, Children's Mercy Hospital, Kansas City, MO

Teaching:

1992- Genetics Laboratory Tutorials: Demonstrator, first year Harvard medical students
1992- Cytogenetics lectures, Beth Israel Hospital: Lecturer, first through fourth year Pathology residents
1992- Cytogenetics Laboratory Rotations: Director on service, students, second year Pathology residents and Genetics Fellows in Harvard Training Program in Genetics, ~5 per year at one month each
1993- Cytogenetic and molecular genetics lectures for fellows in Harvard Training Program in Genetics
1996, 1997 Prenatal Diagnosis Lecture to students in HMS/MIT Reproductive Biology progam
2001- Clinical Cytogenetics, UMKC Medical Students (3rd Year)

Advising Responsibilities:

1994 1 medical student in research laboratory
1992-3 1 postdoctoral fellow in research laboratory
1993-99 1 research technician
1991-99 5 clinical technologists
1992-99 clinical pathology residents rotating through the clinical laboratory
1992-99 Faculty, Harvard Genetics Training program
1993-99 Molecular genetic presentations by medical students at perinatal conference
1995-97 1 medical graduate in research laboratory
1996-98 1 clinical genetics fellow in research laboratory
1996-99 visiting students, fellows in genetics
1999- 14 clinical technologists, 2 office assistants
1999- 2 research assistants

Bibliography:

Original Reports:

1. Knoll JHM. Frequency and replication pattern of fragile Xq28 in human heterozygotes from families with X-linked mental retardation. University of Saskatchewan, 1982.

2. Knoll JHM. Roberts Syndrome: Cytological and molecular investigations. University of Manitoba, 1987.

3. Chudley AE, Knoll JH, Gerrard JW, Shepel L, McGahey E, Anderson J. Fragile X-linked mental retardation I: Effect of age and intelligence on expression of the fragile X. Amer J Hum Genet 1983; 14:699-712.

4. Knoll JH, Chudley AE, Gerrard JW. Fragile X-linked mental retardation II: Frequency and replication pattern of fragile Xq28 in heterozygotes. Amer J Hum Genet 1984; 36:640-645.

5. Chudley AE, Rozdilsky B, Houston CS, Becker LE, Knoll JH. Multicore disease in sibs with severe mental retardation, short stature, facial anomalies, hypoplasia of the pituitary fossa, and hypogonadatropic hypogonadism. Amer J Med Genet 1985; 20:145-158.

6. Hagerman RJ, Chudley AE, Knoll JH, Jackson AW, Kemper M, Ahmad R. Autism in fragile X females. Amer J Med Genet 1986; 23:375-380.

7. Heartlein MW, Knoll JHM, Latt SA. Chromosome instability associated with human alphoid DNA transfected into the Chinese hamster genome. Mol Cell Biol 1988; 8(9):3611-3618.

8. Knoll JHM, Nicholls RD, Magenis E, Graham JM Jr, Lalande M, Latt SA. Angelman and Prader-Willi syndromes share a common chromosome 15 deletion but differ in parental origin of the deletion. Amer J Med Genet 1989; 32(2):285-290.

9. Nicholls RD, Knoll JH, Glatt K, Hersh JH, Brewster TD, Graham JM Jr, Wurster-Hill D, Wharton R, Latt SA. Restriction fragment length polymorphisms within proximal 15q and their use in molecular cytogenetics and the Prader-Willi syndrome. Amer J Med Genet 1989; 33:66-77.

10. Knoll JHM, Nicholls RD, Lalande M. On the parental origin of chromosome 15q11q13 in Angelman Syndrome. Hum Genet 1989; 83:205-206.

11. Nicholls RD, Knoll JHM, Butler MG, Karam S, Lalande M. Genetic imprinting suggested by maternal heterodisomy in nondeletion Prader-Willi syndrome. Nature 1989; 342:281-285.

12. Knoll JHM, Nicholls RD, Magenis RE, Glatt K, Graham JM Jr, Kaplan L, Lalande M. Angelman Syndrome: Three molecular classes identified with chromosome 15q11q13 specific DNA markers. Amer J Hum Genet 1990; 47:149-155.

13. Bianchi DW, Flint AF, Pizzimenti MF, Knoll JHM, Latt SA. Isolation of fetal DNA from nucleated erythrocytes in maternal blood. Proc Nat Acad Sci 1990; 87(9):3279-3283.

14. Knoll JHM, Glatt K, Nicholls RD, Malcolm S, Lalande M. Chromosome 15 uniparental disomy is not frequent in Angelman Syndrome. Amer J Hum Genet 1991; 48:16-21.

15. Wagstaff J, Knoll JHM, Fleming J, Kirkness EF, Martin-Gallardo A, Greenberg F, Graham JM Jr, Menninger J, Ward D, Venter JC, Lalande M. Localization of the gene encoding the $GABA_A$ receptor B3 subunit to the Angelman/Prader-Willi region of human chromosome 15. Amer J Hum Genet 1991; 49:330-337.

16. Chaillet JR, Knoll JHM, Horsthemke B, Lalande M. The syntenic relationship between the critical deletion region for the Prader-Willi/Angelman syndromes and proximal mouse chromosome 7. Genomics 1991; 11:773-776.

17. Buiting K, Greger V, Horstmann I, Ludecke JJ, Senger G, Claussen U, Brownstein BH, Schlessinger D, Knoll JHM, Lalande M, Zabel B, Horsthemke B. Microdissection and molecular analysis of proximal 15q. In: Cassidy SB, ed. Prader-Willi syndrome and other chromosome 15q deletion disorders. Springer Verlag, Berlin. Series H: Cell Biology 1991; 61:13-17.

18. Beggs AH, Byers TJ, Knoll JHM, Boyce FM, Bruns G, Kunkel LM. Cloning and characterization of two human skeletal muscle alpha-actinin genes on chromosomes one and eleven. J Biol Chem 1992; 267:9281-9288.

19. Wagstaff J, Knoll JHM, Glatt KA, Shugart YY, Sommer A, Lalande M. Linkage of nondeletion Angelman syndrome to chromosome 1511-q13: Maternal but not paternal transmission leads to phenotypic expression. Nature Genetics 1992; 1:291-294.

20. Webb T, Clayton-Smith J, Cheng X-J, Knoll JHM, Lalande M, Pembrey ME, Malcolm S. Angelman syndrome with a chromosomal inversion 15 inv(p11q13) accompanied by a deletion in 15q11q13. J Med Genet 1992; 29:921-924.

21. Knoll JHM, Wagstaff J, Lalande M. Cytogenetic and Molecular Studies in Prader-Willi and Angelman Syndromes. Amer J Med Genet 1993; 17:694-698.

22. Knoll JHM, Sinnett D, Wagstaff J, Glatt K, Wilcox AS, Whiting P, Wingrove P, Sikela JM, Lalande M. FISH ordering of reference markers and of the gene for the □5 subunit of the gamma-aminobutyric acid receptor (GABRA5) within the Angelman and Prader-Willi syndrome chromosome regions. Hum Molec Genet 1993; 2:183-189.

23. Warman ML, Tiller GE, Polumbo PA, Seldin MF, Rochelle JM, Knoll JHM, Cheng SD, Olsen BR. Physical and linkage mapping of the human and murine genes for the □1 chain of type IX collagen (COL9A1). Genomics 1993; 17:694-698.

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26. Knoll JHM, Cheng SD, Lalande M. Allele specificity of DNA replication timing in the Angelman/Prader-Willi syndrome imprinted chromosomal region. Nature Genet 1994; 6:41-46.

27. Warman ML, McCarthy MT, Peralla M, Vubrio E, Knoll JHM, McDaniels CN, Mayne R, Beier DR, Olsen BR. The genes encoding alpha 2 9 collagen (Col 9A2) map to human chromosome 1p32.3 to 1p33 and mouse chromosome 4. Genomics 1994; 23:158-162.

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31. Mundlos S, Mulliken JB, Abramson DL, Warman ML, Knoll JHM, Olsen BR. Genetic mapping of celeiodocranial dysplasia and evidence of a microdeletion in one family. Hum Molec Genet 1995; 4:71-75.

32. White L, Knoll JHM. Angelman Syndrome: Routine molecular cytogenetic analysis of chromosome 15q11-q13. Amer J Med Genet 1995; 56:101-105.

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34. Saal K, Knoll JHM, Kadin M. Cytogenetic findings in regressing skin lesions of lymphomatoid papulosis. Canc Genet & Cytogenet 1995; 80:13-16.

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39. Schwartz F, Eisenman R, Knoll J, Gessler M, Bruns G. cDNA sequence, genomic organization and evolutionary conservation of a novel gene from the WAGR region. Genomics 1995; 29(2):526-532.

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51. Repetto GR, Korf BR, Wagstaff J, Knoll JHM. Complex familial rearrangement of chromosome 9p24 detected by FISH. Amer J Med Genet, 1998; 76:306-309.

52. Blank V, Knoll JHM, Andrews NC. Molecular characterization and localization of the human MAFG gene. Genomics 1997; 44:147-149.

53. Kim AC, Peters LL, Knoll JHM, VanHuffel, Ciciotte SL, Kleyn P, Chisti AH. Limatin (LIMAB1), an actin-binding LIM protein, maps to mouse chromosome 19 and human chromosome 10q25, a region frequently deleted in human cancers. Genomics 1997; 46:291-293.

54. Tiller GE, Warman ML, Gong Y, Knoll JHM, Mayne R, Brewton RG. Physical and linkage mapping of the gene for the alpha 3 chain of type IX collagen, COL9A3, to human chromosome 20q13.3. Cell Genetics and Cytogenetics 1998; in press.

55. Ott G, Katzenberger T, Siebert R, DeCoteau JF, Fletcher JA, Knoll JHM, Kalla J, Rosenwald A, Michaelason M, Ott MM, Weber-Matthiesen K, Kadin ME, Muller-Hermelink HK. Chromosomal abnormalities in nodal and extranodal CD30+ anaplastic large cell lymphomas: Infrequent detection of the t(2;5) in extranodal lymphomas. Genes, Chromosomes and Cancer 1998; 22:114-121.

56. Rogan PK, Seip JR, White LM, Wenger SL, Steele MW, Sperling MA, Menon R, Knoll JHM. Relaxation of imprinting in Prader-Willi syndrome. Human Genetics 1998: 103:694-701.

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58. Repetto GM, White LM, Bader PJ, Johnson D, Knoll JHM. Interstitial duplication of proximal chromosome 15q in three patients with autistic features and mental retardation. Amer J Med Genet 1998; 79:82-89.

59.Hahn WC, Stewart SA, Brookls M, York SG, Ng-Eaton E, Kurachi A, Beijersbergen RL, Knoll JHM, Meyerson M, Weinberg RA. Inhibition of tilomerase limits the growth of human cancer cells. Nature Medicine 1999; 5:1164-1170.

60. Kocher O, Comella N, Gilchrist A, Pal R, Tognazzi K, Brown LF, Knoll JHM. PDZK, a novel PDZ domain-containing protein upregulated in carcinomas and mapped to chromosome 1q21, interacts with cMOAT (MRPs), the multidrug resistance-associated protein. Lab Invest 1999; 79(9):1161-70 (issue cover).

61. Austin-Ward ED, Castillo S, Gragnic Y, Sanz P, Salazar S, Knoll JHM. Clinical findings in a patient with a supernumerary ring chromosome 20. Amer J Med Genet 91:171-174, 2000.

62. Ming JE, Blagowidow N, Knoll JH, Rollings L, Fortina P, McDonald-McGinn DM, Spinner NB, Zackai EH. A submicroscopic deletion in cousins with Prader-Willi syndrome causes a grandmatrilineal inheritance pattern: effects of imprinting. Amer J Med Genet 92:19-24, 2000.

63. Rogan, PK, Cazcarro, P, Knoll JHM. Sequence-based design of single-copy genomic DNA probes for fluorescence in situ hybridization. Genome Research 11: 1086-1094, 2001.

64. Kruskall MS, Yu N, Yunis JJ, Knoll J, Uhl L, Clavijo O, Husain Z, Yunis E, Yunis JJ, Yunis EJ. Tetragametic chimerism in a phenotypically normal woman. New England J Med. In press, May 2002.

Reviews and educationally relevant publications:

1. Knoll JHM, Wagstaff J, Lalande M. Prader-Willi Syndrome. In: Smith B, Adelman G, eds. Neuroscience Year: Supplement 3 to the Encyclopedia of Neuroscience. Birkhauser, Boston, MA. 1993; pp 132-133.

2. Lalande M, Wagstaff J, Knoll JHM. Molecular Analysis of the Angelman/Prader-Willi Syndromes. In: Adolph KW, ed. Genome Research in Molecular Medicine and Virology, New York, NY. Academic Press. 1993; pp 69-82.

3. Lalande M, Wagstaff J, Sinnett D, Greger V, Knoll JHM. Mapping of the Angelman and Prader-Willi syndromes. In Epstein CJ (ed): The phenotypic mapping of Down syndrome and other aneuploid conditions, 1993; 225-234, Wiley-Liss, New York.

4. Knoll JHM, Lichter P. In situ hybridization to metaphase chromosomes and interphase nuclei. In Dracopoli NC, Haines JL, Korf BR, Moir DT, Morton CC, Seidman CE, Seidman JG, Smith DR (eds): "Current protocols in Human Genetics Volume 1" 1994: Unit 4.3, Green-Wiley, New York.

5. Knoll JHM. Prader-Willi Syndrome. In: Smith B, Adelman G, Eds. Neuroscience Year: Supplement 4 to the Encyclopedia of Neuroscience. Birkhauser, Boston, MA. 1996.

6. Robinson, WP, Horsthemke B, Leonard S, Malcolm S, Morton CC, Nicholls RD, Ritchie R, Rogan P, Schultz R, Sxhwartz S, Sharp J, Trent R, Wevrick R, Williamson M, Knoll JHM. Report of the third international workshop on human chromosome 15 mapping 1996. Cytogenetics and Cell Genetics 76:1-13, 1997.

7. Knoll JHM. Genomic Imprinting. In: McGraw-Hill 1999 Yearbook of Science and Technology. December, 1998.

8. Knoll JHM. A Cytogenetic nightmare. Case 4 for Genetics, Embryology, and Reproduction for first year medical students, Harvard Medical School, 1997; 1998; 1999.

Abstracts (not complete listing):

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2. Knoll JH, Chudley AE, Gerrard JW. Efficacy of FUdR in enhancing expression of fragile X in affected males and carrier females with fragile X-linked mental retardation. Amer J Hum Genet 1982; 34(6):131A.

3. Knoll JH, Ray M, Davies D. Effects of cocultivation on heterochromatin puffing in Roberts Syndrome. Genetics Society of Canada Meeting (1983), Ottawa, Ontario: A10.

4. Knoll JH, Ray M. Morphology of constitutive heterochromatin in Roberts Syndrome cells. Genetics 1984; 107(3):s57.

5. Knoll JH, Ray M. Roberts Syndrome: Correction of chromosomal abnormality by somatic cell fusion. Genetics Society of Canada Meeting (1986), Quebec City, Quebec: OHG-6.

6. Korneluk RG, Wang HS, MacLeod HL, Knoll JH, Bruns GAP, Adra C, Hunter A. Cloning and sublocalization of the human chromosome 19 phosphoglycerate kinase (PGK) sequences. Cytogenet Cell Genet 1987; 46(1-4):641.

7. Wang HS, Knoll JH, Korneluk RG. Sublocalization of a human autosomal sequence homologous to X-linked phosphoglycerate kinase using in situ hybridization. Amer J Hum Genet 1987; 41(3)s:A189.

8. Treacy E, Vekemans M, Blaichman S, Lavallie S, Polychronakos C, Xu U, Knoll J, Lalande M, Der Kaloustian

V. De novo translocation between chromosomes 6 and 15 and detailed mapping of IGFi/Mannose-6-phosphate receptor. 8th International Congress of Human Genetics. Amer J Hum Genet 1991; 49(4s):272.

9. Williams CA, Angelman H, Clayton-Smith J, Driscoll DJ, Hendrickson JE, Knoll JHM, Magenis RE, Schinzel, Wagstaff J, Whidden EM, Zori R. Angelman syndrome: consensus for diagnostic criteria. Amer J Hum Genet 1994; 55(3):A96.

10. Beggs AH, Arahata K, Watkins SC, Knoll JHM, Kunkel LM. Cloning of a novel human nonmuscle alpha-actinin gene and characterization of alpha-actinin expression in nemaline rod myopathy. Amer J Hum Genet 1994; 55(3):A128.

11. Mundlos S, Knoll JHM, Mulliken JB, Warman ML, Olsen BR. Genetic mapping of cleiodocranial dysplasia and detection of a microdeletion in one family. International Genetic Workshop on Crouzon and Other Craniofacial Disorders, March (1995).

12. White LM, DerKaloustian V, Naeem R, Knoll JHM. Chromosome 15q11q13 duplications: Mechanism of formation. Amer J Hum Genet 1995; 57(4):A130.

13. Knoll JHM, Rogan PK, Nicholls RD, Wu B-L, Korf B, White LM. Allele-specific replication of 15q11q13 loci: A diagnostic test for detection of uniparental disomy. Amer J Hum Genet 1995; 57(4):A34.

14. Rogan PK, Seip JR, Knoll JHM, White LM, Wenger SL, Steele MW, Sperling MA, Menon R. Relaxation of imprinting patients with Prader-Willi syndrome. Amer J Hum Genet (1996); 59(4):A39.

15. Blagowidow N, Ming JE, McDonald-McGinn DM, Speare V, Sovinsky L, Knoll JHM, Zackai EH, Spinner NB. Grandmatrilineal inheritance of a small chromosome 15q11.2 deletion in an extended family with Prader-Willi syndrome. Amer J Hum Genet (1996); 59(4):A165.

16. Mundlos S, Mundlos C, Knoll JHM, Albright S, Aylsworth, A, Henn W, Curry, Olsen BR. Microdeletions in cleidocranial dysplasia. Amer J Hum Genet (1996); 59(4):A274.

17. Knoll JHM, White LM, Repetto G, Der Kaloustian V, Mundlos C, Eydoux P, Korf B. Large supernumerary dicentric chromosome 15q11q13 duplications: Mulitple regions of breakage, mechanisms of formation and clinical findings. Amer J Hum Genet (1997); 61(4):A7.

18. Repetto GM, White LM, Bader PJ, Johnson D, Knoll JHM.
Interstitial duplication of proximal chromosome 15q in three patients with autistic features and mental retardation. Amer J Hum Genet (1997); 61(4):A139.

19. Tiller GE, Warman ML, Gong Y, Knoll JHM, Mayne R, Brewton
RG. Physical and linkage mapping of the gene for the alpha3 chain of type IX collagen, COL9A3, to human chromosome 20q13.3. Amer J Hum Genet (1997); 61(4):A297.

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21. JHM Knoll, H Baker, G Cox, ML Begleiter, LM Pasztor. Parental origin and replication timing studies in a 70,XXXX liveborn. Amer J Hum Genet 65(4):A167.

22. JHM Knoll. Molecular Cytogenetics as an Adjunct to Conventional Chromosome Analysis in Cancer. Greater KC. Cancer Research Group, April 2000, Stowers Institute, Kansas City.

23. Knoll JHM, Cazcarro P, Rogan PK. Clinical application of sequence-based single copy probes for FISH.

Amer J Hum Genet 67(4):A157, 2000.

24. Mouron B, Begleiter ML, Rinaldi R, Butler MG, Knoll JHM. Inherited chromosome Xp22.3 deletion: discordant phenotype in mother and daughter. Amer J Hum Genet 67(4):A161, 2000.

25. Rogan PK, Cazcarro P, Knoll JHM. Single copy hybridization probes derived by genomic sequence analysis. Amer J Hum Genet 67(4):A267, 2000.